

CLAIMS

1. A nucleic acid molecule comprising a chromosomal region contributing to or indicative of hyperlipidemias and/or dyslipidemias and/or defective carbohydrate metabolism, wherein said nucleic acid molecule is selected from the group consisting of:
 - (a) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence has one or more mutations having an effect on USF1 function;
 - (b) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence is characterized by comprising a guanine or an adenine residue in position 3966 in intron 7 of the USF1 sequence; and/or
 - (c) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, wherein said nucleic acid sequence is characterized by comprising a cytosine or a thymine residue in position 5205 in exon 11 of the USF1 sequence;wherein said nucleic molecule extends, at a maximum, 50000 nucleotides over the 5' and/or 3' end of the nucleic acid molecule of SEQ ID NO: 1.
2. The nucleic acid molecule of claim 1 which is genomic DNA.
3. A fragment of the nucleic acid molecule of claim 1 or 2 having at least 20 nucleotides wherein said fragment comprises nucleotide position 3966 and/or position 5205 of SEQ ID NO:1.
4. A nucleic acid molecule which is complementary to the nucleic acid molecule of any one of claims 1 to 3 and which has a length of at least 20 nucleotides.
5. A vector comprising the nucleic acid molecule of any one of claim 1 to 4.

6. A primer or primer pair, wherein the primer or primer pair hybridizes under stringent conditions to the nucleic acid molecule of any one of claims 1 to 4 comprising nucleotide positions 3966 and 5205 SEQ ID NO:1 or to the complementary strand thereof.
7. A non-human host transformed with the vector of claim 5.
8. The non-human host of claim 7 which is a bacterium, a yeast cell, an insect cell, a fungal cell, a mammalian cell, a plant cell, a transgenic animal or a transgenic plant.
9. A pharmaceutical composition comprising USF1 or a fragment thereof, a nucleic acid molecule encoding USF1 or a fragment thereof, or an antibody specific for USF1.
10. A diagnostic composition comprising a nucleic acid molecule encoding USF1 or a fragment thereof, the nucleic acid molecule of any one of claims 1 to 4, the vector of claim 5, the primer or primer pair of claim 6 or an antibody specific for USF1.
11. A method for testing for the presence or predisposition of hyperlipidemia and/or dyslipidemia and/or defective carbohydrate metabolism, comprising analyzing a sample obtained from a prospective patient or from a person suspected of carrying such a predisposition for the presence of a wild-type or variant allele of the USF1 gene.
12. The method of claim 11, wherein said variant comprises an SNP at position 3966 and/or at position 5205 of the USF1 gene in a homozygous or heterozygous state.
13. The method of claim 11 or 12, wherein said testing comprises hybridizing the complementary nucleic acid molecule of claim 4 under stringent conditions to nucleic acid molecules comprised in a sample and detecting said hybridization,

wherein said complementary nucleic acid molecule comprises the sequence position containing the SNP.

14. The method of any one of claim 11 to 13 further comprising digesting the product of said hybridization with a restriction endonuclease or subjecting the product of said hybridization to digestion with a restriction endonuclease and analyzing the product of said digestion.
15. The method of claim 14, wherein said probe is detectably labeled.
16. The method of any one of claims 11 to 15, wherein said testing comprises determining the nucleic acid sequence of at least a portion of the nucleic acid molecule of any one of claims 1 to 4, wherein said portion comprises the position of the SNP.
17. The method of claim 16, wherein the determination of the nucleic acid sequence is effected by solid-phase minisequencing.
18. The method of claim 17 further comprising, prior to determining said nucleic acid sequence, amplification of at least said portion of said nucleic acid molecule.
19. The method of claim 11 to 15, wherein said testing comprises carrying out an amplification reaction wherein at least one of the primers employed in said amplification reaction is the primer of claim 6 or belongs to the primer pair of claim 6, comprising assaying for an amplification product.
20. The method of claim 19 wherein said amplification is effected by or said amplification is the polymerase chain reaction (PCR).
21. A method for testing for the presence or predisposition of hyperlipidemia and/or dyslipidemia and/or defective carbohydrate metabolism comprising assaying a sample obtained from a human for the amount of (a) USF1, (b) ABCA1, (c) angiotensinogen or (d) apolipoprotein E contained in said sample.

22. The method of claim 21, wherein said testing is effected by using an antibody or aptamer specific for (a) USF1, (b) ABCA1, (c) angiotensinogen or (d) apolipoprotein E.
23. The method of claim 22, wherein said antibody or aptamer is detectably labeled.
24. The method of any one of claims 21 to 23, wherein the test is an immunoassay.
25. A method for testing for the presence or predisposition of hyperlipidemia and/or dyslipidemia and/or defective carbohydrate metabolism comprising assaying a sample obtained from a human for the amount of RNA encoding (a) ABCA1, (b) angiotensinogen or (c) apolipoprotein E contained in said sample.
26. The method of any one of claims 11 to 25, wherein said sample is blood, serum, plasma, fetal tissue, saliva, urine, mucosal tissue, mucus, vaginal tissue, fetal tissue obtained from the vagina, skin, hair, hair follicle or another human tissue.
27. The method of any one of claims 11 to 26, wherein the nucleic acid molecule or protein from said sample is fixed to a solid support.
28. The method of claim 27, wherein said solid support is a chip, a silica wafer, a bead or a microtiter plate.
29. Use of the nucleic acid molecule of any one of claims 1 to 5 for the analysis of the presence or predisposition of hyperlipidemia and/or dyslipidemia and/or defective carbohydrate metabolism.
30. Use of USF1 or a fragment thereof or of a nucleic acid molecule encoding USF1 and/or comprising at least the wild-type sequence of intron 7 and/or exon 11 of USF1, for the preparation of a pharmaceutical composition for the treatment of hyperlipidemias and/or dyslipidemias including familial combined

hyperlipidemia (FCHL), hypercholesterolemia, hypertriglyceridemia, hypoalphalipoproteinemia, hyperapobetalipoproteinemia (hyperapoB), familial dyslipidemic hypertension (FDH), metabolic syndrome, type 2 diabetes mellitus, coronary heart disease, atherosclerosis or hypertension.

31. Kit comprising the nucleic acid molecule of any one of claims 1 to 5, the primer or primer pair of claim 6 and/or the vector of claim 7 in one or more containers.
32. Use of an inhibitor of expression of USF1, wherein said inhibitor is (a) an siRNA or antisense RNA molecule comprising a nucleotide sequence complementary to the transcribed region of the USF1 gene or (b) of an antibody, aptamer or small inhibitory molecule specific for USF1, for the preparation of a pharmaceutical composition for the treatment of hyperlipidemias and/or dyslipidemias including familial combined hyperlipidemia (FCHL), hypercholesterolemia, hypertriglyceridemia, hypoalphalipoproteinemia, hyperapobetalipoproteinemia (hyperapoB), familial dyslipidemic hypertension (FDH), metabolic syndrome, type 2 diabetes mellitus, coronary heart disease, atherosclerosis or hypertension.
33. Use of an activator of expression of USF1 for the preparation of a pharmaceutical composition for the treatment of hyperlipidemias and/or dyslipidemias including familial combined hyperlipidemia (FCHL), hypercholesterolemia, hypertriglyceridemia, hypoalphalipoproteinemia, hyperapobetalipoproteinemia (hyperapoB), familial dyslipidemic hypertension (FDH), metabolic syndrome, type 2 diabetes mellitus, coronary heart disease, atherosclerosis or hypertension, wherein said activator is a small molecule.